



## ALOXE3 gene

arachidonate lipoxygenase 3

### Normal Function

The *ALOXE3* gene provides instructions for making an enzyme called eLOX3. This enzyme is part of a family of enzymes called lipoxygenases. Most enzymes in this family help add an oxygen molecule to certain fatty acids to produce substances called fatty acid hydroperoxides.

Unlike other lipoxygenases, the eLOX3 enzyme does not act directly on fatty acids. Instead, it processes the product of another lipoxygenase reaction, a fatty acid hydroperoxide. The substance produced is later converted to a signaling molecule that is involved in the growth and division (proliferation) and specialization (differentiation) of skin cells.

The eLOX3 enzyme is thought to play a role in the formation and maintenance of the fat (lipid) membrane of the cells that make up the outermost layer of the skin (the epidermis). The epidermis helps prevent water loss, regulates body temperature, and protects against infection.

### Health Conditions Related to Genetic Changes

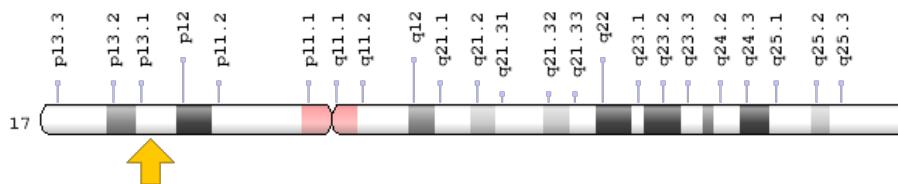
#### nonbullous congenital ichthyosiform erythroderma

At least 10 mutations in the *ALOXE3* gene have been found to cause nonbullous congenital ichthyosiform erythroderma (NBCIE). Most of these mutations change single protein building blocks (amino acids) in the eLOX3 enzyme. Many *ALOXE3* gene mutations lead to the production of a nonfunctional eLOX3 enzyme, which impairs the formation of the lipid membrane of cells within the epidermis. Problems with this protective barrier underlie the skin abnormalities and other features of NBCIE.

## Chromosomal Location

Cytogenetic Location: 17p13.1, which is the short (p) arm of chromosome 17 at position 13.1

Molecular Location: base pairs 8,095,900 to 8,118,916 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- E-LOX
- e-LOX-3
- eLOX3
- epidermal lipoxygenase
- LOXE3\_HUMAN
- MGC119694
- MGC119695
- MGC119696

## Additional Information & Resources

### GeneReviews

- Autosomal Recessive Congenital Ichthyosis  
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28ALOXE3%5BTIAB%5D%29+OR+%28eLOX3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

## OMIM

- ARACHIDONATE LIPOXYGENASE 3  
<http://omim.org/entry/607206>

## Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=ALOXE3%5Bgene%5D>
- HGNC Gene Family: Arachidonate lipoxygenases  
<http://www.genenames.org/cgi-bin/genefamilies/set/407>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=13743](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=13743)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/59344>
- UniProt  
<http://www.uniprot.org/uniprot/Q9BYJ1>

## **Sources for This Summary**

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